

Illinois Department of Public Health  
Newborn Screening Laboratory Subcommittee  
Illinois Department of Public Health, Division of Laboratories  
2121 W. Taylor St., Chicago, Illinois  
Meeting and Conference Call Minutes: December 3, 2014

Subcommittee Members Attending:

George Hoganson-University of Illinois at Chicago – Chair  
Denise Lonigro-Advocate Christ Hospital  
W. Patrick Zeller- Endocrinologist- Private Practice  
Gopal Srinivasan-Mt. Sinai Hospital  
Ramsay Fuleihan- Lurie Children’s Hospital  
Barbara Burton-Lurie Children’s Hospital  
Kristin Clemenz-Lurie Children’ Hospital

IDPH Staff:

George Dizikes, Matt Charles, Arthur Kohrman, Khaja Basheeruddin, Raj Singh, John Nawrocki, Bill Calvert, Jennifer Crew, Joel Price, Hector Diaz, Tom Schafer, Claudia Nash, Shannon Harrison, Jean Becker, Heather Shryock, Nitika Sharma

The meeting was called to order at 9:05 AM, followed by introductions. The minutes were approved for the meeting held September 17, 2014.

**Old Business**

There were no items for discussion.

**New Business**

**SCID Testing- Six Month Update**

Data were reviewed for the six month period of statewide SCID testing and for the pilot test period. During this time frame there were 341 abnormal tests on preterm infants and 150 on term infants with 2 cases of SCID diagnosed and 4 newborns with DiGeorge syndrome. The IDPH Follow-Up Program staff have received several comments from physicians about the high number of false positives being reported and the difference in our cutoff compared to the neighboring state of Wisconsin. In response to this issue, staff from the IDPH Newborn Screening Lab and Follow-Up Program had a discussion with staff from the Massachusetts Department of Public Health, which uses a similar testing methodology as Illinois. Dr. Dizikes reported that the Massachusetts Newborn Screening Program has approximately one-fifth the number of cases compared to Illinois referred for flow cytometry, but has not diagnosed as many positive cases as Illinois, even with a longer history of testing. IDPH lab staff will compare proficiency testing results with Massachusetts and will continue to review screening data and may consider changing the cutoff to 250 TRECS . It was suggested that more information be disseminated to primary care providers about the importance of following the IDPH recommendations when a positive screen is reported. IDPH staff will work with the Illinois Chapter of the American Academy of Pediatrics to encourage primary care providers to follow the AAP recommendations, and the Illinois Academy of Family Physicians to disseminate more information about SCID newborn screening and the importance of follow-up .

**Lysosomal Storage Disorders- Review of Pilot Test Implementation**

The LSD pilot began November 3, 2014, with newborns from four hospitals being tested for five LSDs; Pompe, Gaucher, Fabry, MPS I and Niemann-Pick. To date, 1,200 samples have been tested with two being positive for Pompe and one sample from a baby in the NICU abnormal for two LSDs. All three newborns are being followed and tested further. The target date for the Krabbe testing pilot is planned for the first two to three months of 2015. Hospitals included in the current pilot phase are Advocate Christ Medical Center, Central DuPage Hospital, OSF St. Francis Hospital in Peoria and the University of Chicago with Prentice/Northwestern to be added as soon as issues are resolved regarding HL7 messaging of test results. The IDPH lab is planning to add other hospitals to the pilot prior to implementing statewide screening.

**Holiday/ Saturday Coverage-Protocol Review and Recommendations**

Holiday operations will be implemented in the upcoming year. In prior years, the lab and follow up staff were operational only on the Friday after Thanksgiving since that is always a four day weekend. The laboratory has decided to implement a holiday schedule to assure accessioning and testing of samples is not delayed due to holidays. Protocols from the laboratory and follow-up programs have been developed, along with a calendar of holiday dates for 2015 where the testing will be performed and reported. Newborn screening hospital contacts will be notified of this procedural change. IDPH is also making plans to implement a six day work week, with testing and reporting on Saturdays. The change will require additional funding and staff, but steps are being taken towards this goal. It is unclear whether the anticipated hiring freeze will occur in January with the new administration, which would affect this new policy. The laboratory subcommittee will provide input on which disorders will be included in Saturday testing/reporting as hiring plans move forward. Follow-up staff will develop a detailed reporting algorithm to provide consistency in the steps taken to report abnormal screening results, since many physician offices are closed on Saturday.

**Laboratory Report**

Dr. George Dizikes discussed handouts regarding specimen receipt/testing turn-around time for September- November; with the mean time from collection to IDPH receipt at 1.6 days; from receipt to report of abnormal results at 4.0 days, and from receipt to report of normal results at 9.7 days.

Data comparing August- November of 2013 and 2014 positive numbers for SCAD/IBCD and MCAD were also reviewed which indicate that SCAD positives increased by nearly 50% and MCAD positives decreased by 50% in 2014. A summary of other abnormal test results was also distributed for September- November 2014.

There was no discussion regarding the data presented.

**Follow-up Program Report**

Claudia Nash reported that Tom Schafer, the Deputy Director over the follow- up program, is retiring the end of December, and has always been a strong advocate for newborn screening at IDPH.

Claudia also reported progress is occurring regarding development of the interface with the Illinois Vital Records System. This will allow for import of data into the newborn screening databases (metabolic and hearing) that is collected for the birth certificate, and will provide much more complete and accurate data and will reduce duplicate data entry for birth hospital staff. Testing should occur during the first weeks of 2015.

The meeting adjourned at 10:00 a.m.